Case reports

A new case of proximal 10q partial trisomy

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Abstract

We report on a girl with mild phenotypic abnormalities and duplication of chromosome 10q11→10q22. The similarities to two previously reported cases with an identical chromosomal aberration provide further support for the delineation of this entity as a specific, clinically recognisable syndrome.

Trisomy for a chromosomal segment is often derived from a balanced translocation in one of the parents. In these cases there is a concomitant monosomy for another segment, and the resulting phenotype is influenced by both events. Cases of 'pure' trisomy tend to present more constant manifestations, and enable a more accurate phenotype-karyotype correlation.

We report here on a girl with a direct duplication of the segment 10q11→10q22 and a phenotype almost identical to that of two previously reported cases with the same trisomy.^{1 2}

Case report

The proband is the second child of healthy, unrelated, young parents; her only sib is a normal boy. She was born after an uneventful pregnancy and delivery, birth weight 2400 g. Hypertonicity was noted during the newborn period.

She was referred to the genetics unit because of psychomotor retardation. On examination at 2 years 8 months, height (84 cm), weight (11 kg), and head circumference (45 cm) were all around the 3rd centile. The following abnormalities were noted: slight facial dysmorphism (fig 1) with deep set, small eyes, short

palpebral fissures, telecanthus, strabismus, short philtrum, bow shaped mouth, and mild micrognathia. The ears were slightly low set with a thick helix. She also had two supernumerary nipples on each side, a single flexion crease on both fifth fingers, and mild camptodactyly with abnormality of the flexion creases on both fourth fingers.

Development was delayed from the start: she sat at 20 months, walked at 25 months, and spoke her first words at 24 months. At 32 months, her vocabulary consisted of a few words and she was able to understand only simple orders. Psychometric tests showed mild mental retardation. No other neurological abnormalities were found. A normal EEG was recorded.

CYTOGENETIC STUDIES

Chromosome analysis of 30 GTG banded metaphases from peripheral blood lymphocytes showed a direct duplication of the 10q11→10q22 segment (fig 2). The karyotype is: 46,XX,dir dup(10)(pter→q22::q11→qter).

Figure 1 The proband aged 2 years 9 months.

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Figure 2 Partial GTG banded karyotype from three cells, and schematic representation of the partial duplication of 10q (arrows). Normal homologue is shown on the left of each pair.

Discussion

Fryns *et al*² reported a case of duplication of the 10q11→10q22 segment and they commented on the similarity of their patient to the one previously described by Vogel *et al*,¹ suggesting the existence of a specific, clinically recognisable syndrome related to proximal partial trisomy 10q. Our patient adds further evidence to this suggestion. This is the third report of trisomy for the same chromosomal region, and the phenotype shows mild but specific clinical features, very similar to those previously described (table).

These patients show mild to moderate delay in mental and physical development. Birth weight is low. The face is characteristic, with small, deep set eyes, strabismus, a short, upturned nose, a prominent philtrum, bowed mouth, and some degree of micrognathia. Ocular abnormalities (microphthalmia, coloboma) may also be a feature. The ears show slight but constant dysmorphic features. The neck tends to be short and the limbs slender, and there may be mild skeletal anomalies (rib duplication and equinovarus feet in one patient,¹ polydactyly and scoliosis in the other²).

Our patient also had supernumerary nipples, which were not reported in the other two cases. At present, it is not possible to know whether this is a feature of the syndrome or just a coincidence.

It is worth mentioning that in all three cases the trisomy originated from the same cytogenetic abnormality, that is, a direct (serial) duplication. This type of chromosome aberration is uncommon; the most likely explanation for it seems to be a spontaneous reciprocal translocation during meiosis in either parent.¹ Whether, as these cases suggest, there

Comparison of the three cases of trisomy 10q11→10q22.

	Vogel <i>et al</i> ¹	Fryns <i>et al</i> ²	Present case
<i>General</i>			
Birth weight (g)	2220	2650	2400
Age at reporting (y)	7	16	2
Developmental delay	+	+	+
Growth retardation	+	+	+
<i>Craniofacial</i>			
Microcephaly	+	+	+
Deep set eyes	+	+	+
Microphthalmia/coloboma	—	+	—
Strabismus	—	+	+
Small, upturned nose	+	+	+
Short philtrum	+	—	+
Prominent philtrum	+	+	+
Bowed mouth	+	+	+
High arched palate	+	+	+
Micrognathia (mild)	+	+	+
Thick ear helix	+	+	+
<i>Skeletal/limbs</i>			
Slender hands	+	+	—
Polydactyly	+	—	—
Camptodactyly	—	—	+
Single crease on 5th finger	—	—	+
Other skeletal anomalies	+	+	—

are some chromosomal segments that could be prone to this kind of rearrangement is a question that remains unresolved.

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1 Vogel W, Back E, Imm W. Serial duplication of 10 (q11→q22) in a patient with minor congenital malformations. *Clin Genet* 1978;13:159–63.

2 Fryns JP, Kleczkowska A, Igodt-Ameys L, Van den Berghe H. Proximal duplication of the long arm of chromosome 10 (10q11.2→10q22): a distinct clinical entity. *Clin Genet* 1987;32: 61–5.